

si18432_SEQ

SEQUENCE LISTING

<110> Samsung Electronics Co. Ltd

<120> A variant of HNF-1a gene having novel single nucleotide polymorphism and a variant protein encoded by the same

<160> 25

<170> KopatentIn 1.71

<210> 1

<211> 1896

<212> DNA

<213> Homo sapiens

<220>

<221> CDS

<222> (1)..(1893)

<223> amino acid sequence of HNF-1a

<400> 1	
atg gtt tct aaa ctg agc cag ctg cag acg gag ctc ctg gcg gcc ctg	48
Met Val Ser Lys Leu Ser Gln Leu Gln Thr Glu Leu Leu Ala Ala Leu	
1 5 10 15	
ctc gag tca ggg ctg agc aaa gag gca ctg atc cag gca ctg ggt gag	96
Leu Glu Ser Gly Leu Ser Lys Glu Ala Leu Ile Gln Ala Leu Gly Glu	
20 25 30	
ccg ggg ccc tac ctc ctg gct gga gaa ggc ccc ctg gac aag ggg gag	144
Pro Gly Pro Tyr Leu Leu Ala Gly Glu Gly Pro Leu Asp Lys Gly Glu	
35 40 45	
tcc tgc ggc ggc ggt cga ggg gag ctg gct gag ctg ccc aat ggg ctg	192
Ser Cys Gly Gly Gly Arg Gly Glu Leu Ala Glu Leu Pro Asn Gly Leu	
50 55 60	
ggg gag act cgg ggc tcc gag gac gag acg gac gac gat ggg gaa gac	240
Gly Glu Thr Arg Gly Ser Glu Asp Glu Thr Asp Asp Asp Gly Glu Asp	
65 70 75 80	
ttc acg cca ccc atc ctc aaa gag ctg gag aac ctc agc cct gag gag	288
Phe Thr Pro Pro Ile Leu Lys Glu Leu Glu Asn Leu Ser Pro Glu Glu	
85 90 95	
gcg gcc cac cag aaa gcc gtg gtg gag acc ctt ctg cag gag gac ccg	336
Ala Ala His Gln Lys Ala Val Val Glu Thr Leu Leu Gln Glu Asp Pro	
100 105 110	
tgg cgt gtg gcg aag atg gtc aag tcc tac ctg cag cag cac aac atc	384
Trp Arg Val Ala Lys Met Val Lys Ser Tyr Leu Gln Gln His Asn Ile	
115 120 125	
cca cag cgg gag gtg gtc gat acc act ggc ctc aac cag tcc cac ctg	432
Pro Gln Arg Glu Val Val Asp Thr Thr Gly Leu Asn Gln Ser His Leu	
130 135 140	
tcc caa cac ctc aac aag ggc act ccc atg aag acg cag aag cgg gcc	480
Ser Gln His Leu Asn Lys Gly Thr Pro Met Lys Thr Gln Lys Arg Ala	
145 150 155 160	

si18432_SEQ

gcc	ctg	tac	acc	tgg	tac	gtc	cgc	aag	cag	cga	gag	gtg	gcg	cag	cag	528
Ala	Leu	Tyr	Thr	Trp	Tyr	Val	Arg	Lys	Gln	Arg	Glu	Val	Ala	Gln	Gln	
				165					170					175		
ttc	acc	cat	gca	ggg	cag	gga	ggg	ctg	att	gaa	gag	ccc	aca	ggt	gat	576
Phe	Thr	His	Ala	Gly	Gln	Gly	Gly	Leu	Ile	Glu	Glu	Pro	Thr	Gly	Asp	
			180					185					190			
gag	cta	cca	acc	aag	aag	ggg	cgg	agg	aac	cgt	ttc	aag	tgg	ggc	cca	624
Glu	Leu	Pro	Thr	Lys	Lys	Gly	Arg	Arg	Asn	Arg	Phe	Lys	Trp	Gly	Pro	
		195					200					205				
gca	tcc	cag	cag	atc	ctg	ttc	cag	gcc	tat	gag	agg	cag	aag	aac	cct	672
Ala	Ser	Gln	Gln	Ile	Leu	Phe	Gln	Ala	Tyr	Glu	Arg	Gln	Lys	Asn	Pro	
	210					215					220					
agc	aag	gag	gag	cga	gag	acg	cta	gtg	gag	gag	tgc	aat	agg	gcg	gaa	720
Ser	Lys	Glu	Glu	Arg	Glu	Thr	Leu	Val	Glu	Glu	Cys	Asn	Arg	Ala	Glu	
					230					235					240	
tgc	atc	cag	aga	ggg	gtg	tcc	cca	tca	cag	gca	cag	ggg	ctg	ggc	tcc	768
Cys	Ile	Gln	Arg	Gly	Val	Ser	Pro	Ser	Gln	Ala	Gln	Gly	Leu	Gly	Ser	
				245					250					255		
aac	ctc	gtc	acg	gag	gtg	cgt	gtc	tac	aac	tgg	ttt	gcc	aac	cgg	cgc	816
Asn	Leu	Val	Thr	Glu	Val	Arg	Val	Tyr	Asn	Trp	Phe	Ala	Asn	Arg	Arg	
			260					265					270			
aaa	gaa	gaa	gcc	ttc	cgg	cac	aag	ctg	gcc	atg	gac	acg	tac	agc	ggg	864
Lys	Glu	Glu	Ala	Phe	Arg	His	Lys	Leu	Ala	Met	Asp	Thr	Tyr	Ser	Gly	
			275				280					285				
ccc	ccc	cca	ggg	cca	ggc	ccg	gga	cct	gcg	ctg	ccc	gct	cac	agc	tcc	912
Pro	Pro	Pro	Gly	Pro	Gly	Pro	Gly	Pro	Ala	Leu	Pro	Ala	His	Ser	Ser	
		290				295					300					
cct	ggc	ctg	cct	cca	cct	gcc	ctc	tcc	ccc	agt	aag	gtc	cac	ggt	gtg	960
Pro	Gly	Leu	Pro	Pro	Pro	Ala	Leu	Ser	Pro	Ser	Lys	Val	His	Gly	Val	
					310				315						320	
cgc	tat	gga	cag	cct	gcg	acc	agt	gag	act	gca	gaa	gta	ccc	tca	agc	1008
Arg	Tyr	Gly	Gln	Pro	Ala	Thr	Ser	Glu	Thr	Ala	Glu	Val	Pro	Ser	Ser	
				325					330					335		
agc	ggc	ggt	ccc	tta	gtg	aca	gtg	tct	aca	ccc	ctc	cac	caa	gtg	tcc	1056
Ser	Gly	Gly	Pro	Leu	Val	Thr	Val	Ser	Thr	Pro	Leu	His	Gln	Val	Ser	
			340					345					350			
ccc	acg	ggc	ctg	gag	ccc	agc	cac	agc	ctg	ctg	agt	aca	gaa	gcc	aag	1104
Pro	Thr	Gly	Leu	Glu	Pro	Ser	His	Ser	Leu	Leu	Ser	Thr	Glu	Ala	Lys	
		355					360					365				
ctg	gtc	tca	gca	gct	ggg	ggc	ccc	ctc	ccc	cct	gtc	agc	acc	ctg	aca	1152
Leu	Val	Ser	Ala	Ala	Gly	Gly	Pro	Leu	Pro	Pro	Val	Ser	Thr	Leu	Thr	
					375						380					
gca	ctg	cac	agc	ttg	gag	cag	aca	tcc	cca	ggc	ctc	aac	cag	cag	ccc	1200
Ala	Leu	His	Ser	Leu	Glu	Gln	Thr	Ser	Pro	Gly	Leu	Asn	Gln	Gln	Pro	
					390					395					400	
cag	aac	ctc	atc	atg	gcc	tca	ctt	cct	ggg	gtc	atg	acc	atc	ggg	cct	1248
Gln	Asn	Leu	Ile	Met	Ala	Ser	Leu	Pro	Gly	Val	Met	Thr	Ile	Gly	Pro	
				405					410					415		

si18432_SEQ

ggt gag cct gcc tcc ctg ggt cct acg ttc acc aac aca ggt gcc tcc Gly Glu Pro Ala Ser Leu Gly Pro Thr Phe Thr Asn Thr Gly Ala Ser 420 425 430	1296
acc ctg gtc atc ggc ctg gcc tcc acg cag gca cag agt gtg ccg gtc Thr Leu Val Ile Gly Leu Ala Ser Thr Gln Ala Gln Ser Val Pro Val 435 440 445	1344
atc aac agc atg ggc agc agc ctg acc acc ctg cag ccc gtc cag ttc Ile Asn Ser Met Gly Ser Ser Leu Thr Thr Leu Gln Pro Val Gln Phe 450 455 460	1392
tcc cag ccg ctg cac ccc tcc tac cag cag ccg ctc atg cca cct gtg Ser Gln Pro Leu His Pro Ser Tyr Gln Gln Pro Leu Met Pro Pro Val 465 470 475 480	1440
cag agc cat gtg acc cag aac ccc ttc atg gcc acc atg gct cag ctg Gln Ser His Val Thr Gln Asn Pro Phe Met Ala Thr Met Ala Gln Leu 485 490 495	1488
cag agc ccc cac gcc ctc tac agc cac aag ccc gag gtg gcc cag tac Gln Ser Pro His Ala Leu Tyr Ser His Lys Pro Glu Val Ala Gln Tyr 500 505 510	1536
acc cac acg ggc ctg ctc ccg cag act atg ctc atc acc gac acc acc Thr His Thr Gly Leu Leu Pro Gln Thr Met Leu Ile Thr Asp Thr Thr 515 520 525	1584
aac ctg agc gcc ctg gcc agc ctc acg ccc acc aag cag gtc ttc acc Asn Leu Ser Ala Leu Ala Ser Leu Thr Pro Thr Lys Gln Val Phe Thr 530 535 540	1632
tca gac act gag gcc tcc agt gag tcc ggg ctt cac acg ccg gca tct Ser Asp Thr Glu Ala Ser Ser Glu Ser Gly Leu His Thr Pro Ala Ser 545 550 555 560	1680
cag gcc acc acc ctc cac atc ccc agc cag gac cct gcc ggc atc cag Gln Ala Thr Thr Leu His Ile Pro Ser Gln Asp Pro Ala Gly Ile Gln 565 570 575	1728
cac ctg cag ccg gcc cac cgg ctc agc gcc agc ccc aca gtg tcc tcc His Leu Gln Pro Ala His Arg Leu Ser Ala Ser Pro Thr Val Ser Ser 580 585 590	1776
agc agc ctg gtg ctg tac cag agc tca gac tcc agc aat ggc cag agc Ser Ser Leu Val Leu Tyr Gln Ser Ser Asp Ser Ser Asn Gly Gln Ser 595 600 605	1824
cac ctg ctg cca tcc aac cac agc gtc atc gag acc ttc atc tcc acc His Leu Leu Pro Ser Asn His Ser Val Ile Glu Thr Phe Ile Ser Thr 610 615 620	1872
cag atg gcc tct tcc tcc cag Gln Met Ala Ser Ser Ser Gln 625 630	1896

taa

<210> 2
<211> 631
<212> PRT
<213> Homo sapiens

si18432_SEQ

```

<400> 2
Met Val Ser Lys Leu Ser Gln Leu Gln Thr Glu Leu Leu Ala Ala Leu
 1      5      10      15
Leu Glu Ser Gly Leu Ser Lys Glu Ala Leu Ile Gln Ala Leu Gly Glu
 20      25      30
Pro Gly Pro Tyr Leu Leu Ala Gly Glu Gly Pro Leu Asp Lys Gly Glu
 35      40      45
Ser Cys Gly Gly Gly Arg Gly Glu Leu Ala Glu Leu Pro Asn Gly Leu
 50      55      60
Gly Glu Thr Arg Gly Ser Glu Asp Glu Thr Asp Asp Asp Gly Glu Asp
 65      70      75      80
Phe Thr Pro Pro Ile Leu Lys Glu Leu Glu Asn Leu Ser Pro Glu Glu
 85      90      95
Ala Ala His Gln Lys Ala Val Val Glu Thr Leu Leu Gln Glu Asp Pro
100      105      110
Trp Arg Val Ala Lys Met Val Lys Ser Tyr Leu Gln Gln His Asn Ile
115      120      125
Pro Gln Arg Glu Val Val Asp Thr Thr Gly Leu Asn Gln Ser His Leu
130      135      140
Ser Gln His Leu Asn Lys Gly Thr Pro Met Lys Thr Gln Lys Arg Ala
145      150      155      160
Ala Leu Tyr Thr Trp Tyr Val Arg Lys Gln Arg Glu Val Ala Gln Gln
165      170      175
Phe Thr His Ala Gly Gln Gly Gly Leu Ile Glu Glu Pro Thr Gly Asp
180      185      190
Glu Leu Pro Thr Lys Lys Gly Arg Arg Asn Arg Phe Lys Trp Gly Pro
195      200      205
Ala Ser Gln Gln Ile Leu Phe Gln Ala Tyr Glu Arg Gln Lys Asn Pro
210      215      220
Ser Lys Glu Glu Arg Glu Thr Leu Val Glu Glu Cys Asn Arg Ala Glu
225      230      235      240
Cys Ile Gln Arg Gly Val Ser Pro Ser Gln Ala Gln Gly Leu Gly Ser
245      250      255
Asn Leu Val Thr Glu Val Arg Val Tyr Asn Trp Phe Ala Asn Arg Arg
260      265      270
Lys Glu Glu Ala Phe Arg His Lys Leu Ala Met Asp Thr Tyr Ser Gly
275      280      285
Pro Pro Pro Gly Pro Gly Pro Gly Pro Ala Leu Pro Ala His Ser Ser
290      295      300
Pro Gly Leu Pro Pro Pro Ala Leu Ser Pro Ser Lys Val His Gly Val
305      310      315      320
Arg Tyr Gly Gln Pro Ala Thr Ser Glu Thr Ala Glu Val Pro Ser Ser
325      330      335

```

si18432_SEQ

Ser Gly Gly Pro Leu Val Thr Val Ser Thr Pro Leu His Gln Val Ser
340 345 350
Pro Thr Gly Leu Glu Pro Ser His Ser Leu Leu Ser Thr Glu Ala Lys
355 360 365
Leu Val Ser Ala Ala Gly Gly Pro Leu Pro Pro Val Ser Thr Leu Thr
370 375 380
Ala Leu His Ser Leu Glu Gln Thr Ser Pro Gly Leu Asn Gln Gln Pro
385 390 395 400
Gln Asn Leu Ile Met Ala Ser Leu Pro Gly Val Met Thr Ile Gly Pro
405 410 415
Gly Glu Pro Ala Ser Leu Gly Pro Thr Phe Thr Asn Thr Gly Ala Ser
420 425 430
Thr Leu Val Ile Gly Leu Ala Ser Thr Gln Ala Gln Ser Val Pro Val
435 440 445
Ile Asn Ser Met Gly Ser Ser Leu Thr Thr Leu Gln Pro Val Gln Phe
450 455 460
Ser Gln Pro Leu His Pro Ser Tyr Gln Gln Pro Leu Met Pro Pro Val
465 470 475 480
Gln Ser His Val Thr Gln Asn Pro Phe Met Ala Thr Met Ala Gln Leu
485 490 495
Gln Ser Pro His Ala Leu Tyr Ser His Lys Pro Glu Val Ala Gln Tyr
500 505 510
Thr His Thr Gly Leu Leu Pro Gln Thr Met Leu Ile Thr Asp Thr Thr
515 520 525
Asn Leu Ser Ala Leu Ala Ser Leu Thr Pro Thr Lys Gln Val Phe Thr
530 535 540
Ser Asp Thr Glu Ala Ser Ser Glu Ser Gly Leu His Thr Pro Ala Ser
545 550 555 560
Gln Ala Thr Thr Leu His Ile Pro Ser Gln Asp Pro Ala Gly Ile Gln
565 570 575
His Leu Gln Pro Ala His Arg Leu Ser Ala Ser Pro Thr Val Ser Ser
580 585 590
Ser Ser Leu Val Leu Tyr Gln Ser Ser Asp Ser Ser Asn Gly Gln Ser
595 600 605
His Leu Pro Ser Asn His Ser Val Ile Glu Thr Phe Ile Ser Thr
610 615 620
Gln Met Ala Ser Ser Ser Gln
625 630

<210> 3
<211> 93
<212> DNA
<213> Homo sapiens

si18432_SEQ

<400> 3
gtaaggcca ggcctgctgg ccctcccttg gcctgtgaca gagcccctca cccccacatc 60
ccccgggctc aggaggctgc tctgctcccc cag 93

<210> 4
<211> 41
<212> DNA
<213> Artificial Sequence

<220>
<223> sense primer for amplifying promoter of MODY3 gene

<400> 4
taatacgact cactataggg tggccgtgag catcctctgc c 41

<210> 5
<211> 39
<212> DNA
<213> Artificial Sequence

<220>
<223> antisense primer for amplifying promoter of MODY3 gene

<400> 5
gtaaccctca ctaaaggac gtgggttgcg ttgcctgc 39

<210> 6
<211> 40
<212> DNA
<213> Artificial Sequence

<220>
<223> sense primer for amplifying exon 1 of MODY3 gene

<400> 6
taatacgact cactataggg cgtggccctg tggcagccga 40

<210> 7
<211> 40
<212> DNA
<213> Artificial Sequence

<220>
<223> antisense primer for amplifying exon 1 of MODY3 gene

<400> 7
gtaaccctca ctaaaggag ggctcgtag gagctgaggg 40

<210> 8
<211> 42
<212> DNA
<213> Artificial Sequence

si18432_SEQ

<220>
 <223> sense primer for amplifying exon 2 of MODY3 gene

<400> 8
 taatacgact cactataggg cccttgctga gcagatcccg tc 42

<210> 9
 <211> 40
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> antisense primer for amplifying exon 2 of MODY3 gene

<400> 9
 gtaaccctca ctaaaggag ggatggtgaa gcttccagcc 40

<210> 10
 <211> 40
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> sense primer for amplifying exon 3 of MODY3 gene

<400> 10
 taatacgact cactataggg gcaaggctag gggaatggac 40

<210> 11
 <211> 42
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> antisense primer for amplifying exon 3 of MODY3 gene

<400> 11
 gtaaccctca ctaaaggag gccgttgtag ctattgcact cc 42

<210> 12
 <211> 43
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> sense primer for amplifying exon 4 of MODY3 gene

<400> 12
 taatacgact cactataggg ggctcatggg tggctatttc tgc 43

<210> 13
 <211> 42

si18432_SEQ

<212> DNA
 <213> Artificial Sequence

<220>
 <223> antisense primer for amplifying exon 4 of MODY3 gene

<400> 13
 gtaaccctca ctaaagggac gtgtcccttg tccccacata cc 42

<210> 14
 <211> 42
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> sense primer for amplifying exon 5 of MODY3 gene

<400> 14
 taatacgact cactataggg tgctgaggca ggacactgct tc 42

<210> 15
 <211> 42
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> antisense primer for amplifying exon 5 of MODY3 gene

<400> 15
 gtaaccctca ctaaagggat acaagcaagg acactcacca gc 42

<210> 16
 <211> 41
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> sense primer for amplifying exon 6 of MODY3 gene

<400> 16
 taatacgact cactataggg cccggacaca gcttggttc c 41

<210> 17
 <211> 42
 <212> DNA
 <213> Artificial Sequence

<220>
 <223> antisense primer for amplifying exon 6 of MODY3 gene

<400> 17
 gtaaccctca ctaaaggga tccccaccag cttaccgatg ac 42

si18432_SEQ

<210> 18
 <211> 40
 <212> DNA
 <213> Artificial Sequence

 <220>
 <223> sense primer for amplifying exon 7 of MODY3 gene

 <400> 18
 taatacgact cactataggg caggcctggc ctccacgcag 40

 <210> 19
 <211> 40
 <212> DNA
 <213> Artificial Sequence

 <220>
 <223> antisense primer for amplifying exon 7 of MODY3 gene

 <400> 19
 gtaaccctca cttaaaggag gggctctgca gctgagccat 40

 <210> 20
 <211> 41
 <212> DNA
 <213> Artificial Sequence

 <220>
 <223> sense primer for amplifying exon 8 and 9 of MODY3 gene

 <400> 20
 taatacgact cactataggg ggcccagtag acccacacgg g 41

 <210> 21
 <211> 40
 <212> DNA
 <213> Artificial Sequence

 <220>
 <223> antisense primer for amplifying exon 8 and 9 of MODY3 gene

 <400> 21
 gtaaccctca cttaaaggag ggcagggaca gtaaggagg 40

 <210> 22
 <211> 41
 <212> DNA
 <213> Artificial Sequence

 <220>
 <223> sense primer for amplifying exon 10 of MODY3 gene

 <400> 22
 taatacgact cactataggg gccttggttg cctctgcagt g 41
 Page 9

si18432_SEQ

<210>	23	
<211>	41	
<212>	DNA	
<213>	Artificial Sequence	
<220>		
<223>	antisense primer for amplifying exon 10 of MODY3 gene	
<400>	23	
gtaaccctca ctaaaggag gccatctggg tggagatgaa g		41
<210>	24	
<211>	20	
<212>	DNA	
<213>	Artificial Sequence	
<220>		
<223>	T7 promoter sequence	
<400>	24	
taatacgact cactataggg		20
<210>	25	
<211>	19	
<212>	DNA	
<213>	Artificial Sequence	
<220>		
<223>	T3 promoter sequence	
<400>	25	
gtaaccctca ctaaaggga		19